

# Human Heredity

## Chapter 14



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### Human Chromosomes

*Humans have 46 Chromosomes- 23 pairs*

**Karyotype** – a picture of arranged chromosomes. The picture shows homologous pairs of autosomes and sex chromosome arranged in order by size.

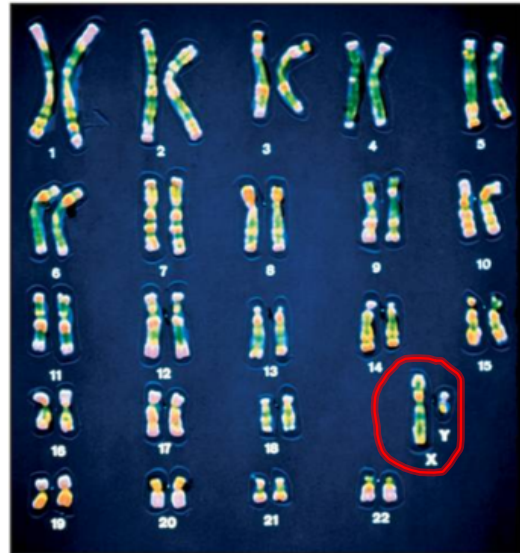


Organism	Number of chromosomes
pea plant	14
sun flower	34
cat	38
puffer fish	42
human	46
dog	78

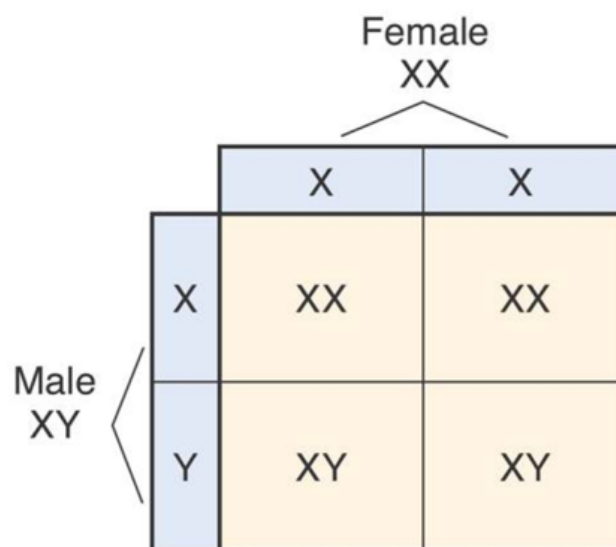
Two of the 46 human chromosomes are known as **sex chromosomes**, because they determine an individual's sex.

Females have two copies of an X chromosome (XX).  
Males have one X chromosome and one Y chromosome(XY).

The remaining 44 chromosomes are known as autosomal chromosomes, or **autosomes**.



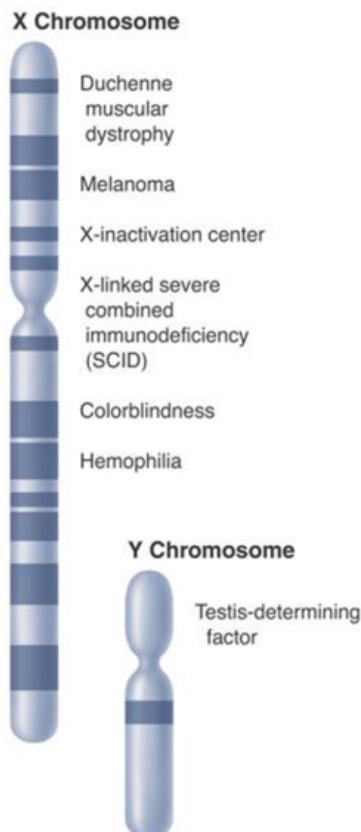
Males and females are born in a roughly 50 : 50 ratio because of the way in which sex chromosomes segregate during meiosis.



# Sex-Linked Genes

Genes located on the X or Y chromosomes are called **sex-linked genes**.

There are more than 100 disorders that have been mapped on the X chromosome. Many of these are recessive traits. Because men only have one X chromosome, these disorders are more common in men.



The Y chromosome is much smaller than the X chromosome and appears to contain only a few genes.

# Sex-Linked Genes

## Hemophilia

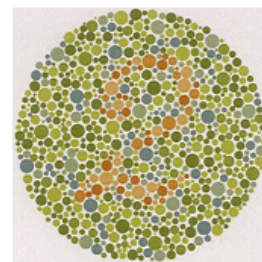
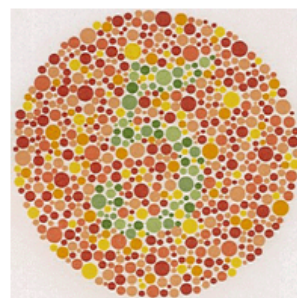
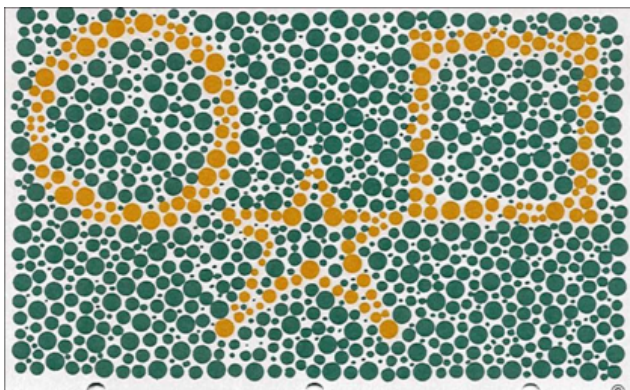
In hemophilia, a protein necessary for normal blood clotting is missing.

The defective gene is on the X chromosome, so males are usually victims and females are usually carriers.

## Color Blindness

The gene for color blindness is recessive and is located on the X chromosome

### Color blindness tests

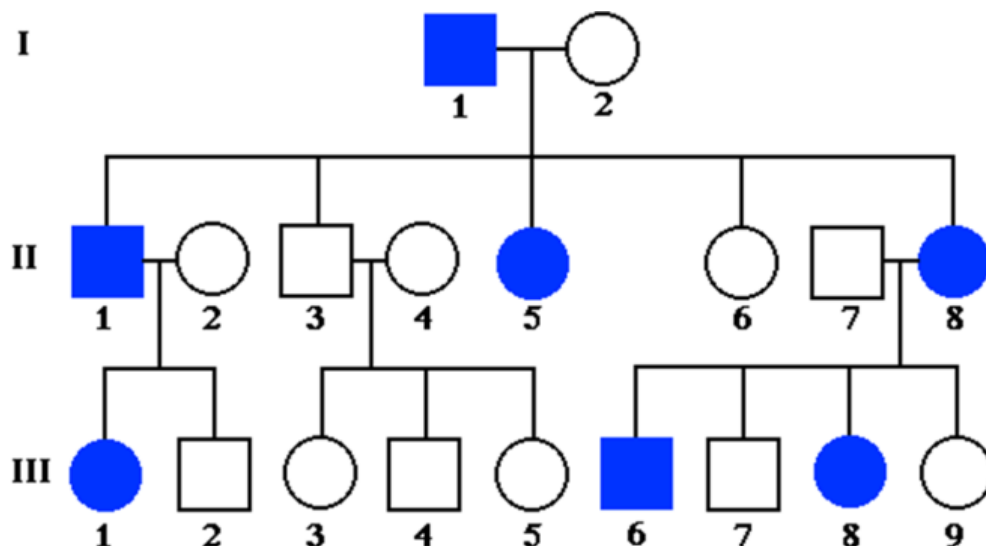
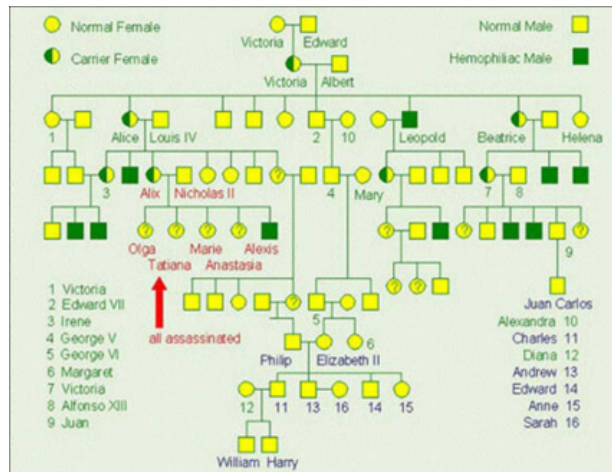


# Human Pedigree

One can study how a trait is passed from one generation to the next. A **pedigree chart**, which shows the relationship within a family of a single trait.

One can infer the genotypes of family members based on when and how a trait is passed from one generation to the next.

A pedigree is not useful when dealing with polygenic traits or some genes that are influenced by nature.



**Pedigree 1. An idealized pedigree of a family with hypercholesterolemia, an autosomal dominant disease where the heterozygote has a reduced number of functional low density lipoprotein receptors.**

# Chromosomal Disorders

The most common error in meiosis occurs when homologous chromosomes fail to separate.

This is known as **nondisjunction**, which means, “not coming apart.”

## Example

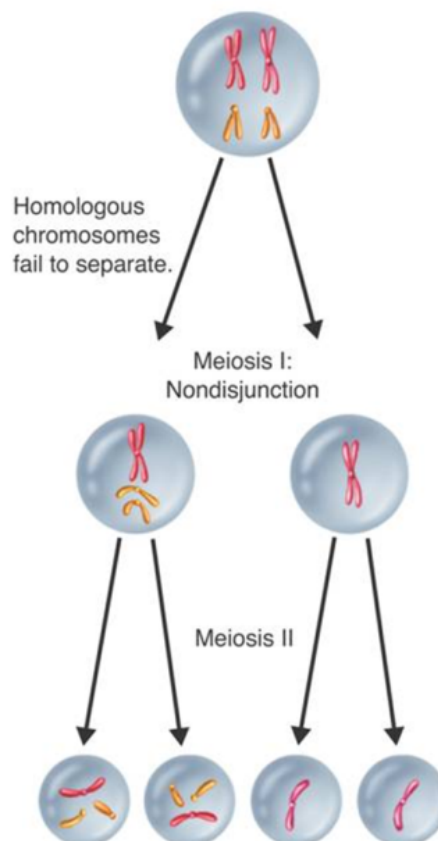
Down Syndrome – 3 copies of chromosome 21. (Trisomy 21)

Sex Chromosomal disorder – improper number of either X or Y chromosomes.

Turner’s syndrome – only one X chromosome.

Klinefelter’s – an extra X chromosomes XXY.

## Nondisjunction





# Single gene genetic disorders

Disorders that are caused when two alleles contribute to the phenotype.

## Example

Sickle Cell Disease – red blood cells twisted and bent.

In sickle cell disease, a small change in the DNA of a single amino acid affects the structure of the protein hemoglobin, causing a serious genetic disorder.

## Human Molecular Genetics

**DNA fingerprinting** – DNA is unique to each person, so it can be used to identify individuals.

